

very severe to milder forms depending on the proportion of cytogenetically normal and trisomic cells. Several major congenital anomalies are usually associated with trisomy 13, including capillary hemangiomas. We report on an 18 month old girl with unexpectedly mild clinical developmental delay presenting with several hemangiomas of different shapes and sizes, but no other malformations.

#### Case report.

2nd child of a 44-year old mother; combined test results in favour of normal fetal development. Dystrophic birth at 28 weeks of gestation, 534 g/33 cm/22 cm (all < Pc3). No cardiopulmonary problems, good muscle tonus, mobility, absolute strength of the extremities and reflexes within normal range of age. Port wine stains in the frontal region; in addition a 4 cm<sup>2</sup> large hemangioma planotuberosum on the forearm. MRI of the brain and eye examination were normal. Due to unspecific dysmorphic features genetic investigation was performed: molecular karyotyping (CytoScan HD, Affymetrix) revealed mosaicism 47,XX,+13/46,XX with 24 % of cells being trisomic at the time of investigation, cytogenetically confirmed with a comparable proportion of mosaicism. Development has been quite promising so far, with weight and length gain along the 3rd percentile. Clinical diagnosis of trisomy 13 is usually straightforward due to specific facial features and malformation pattern, including cardiac and CNS midline defects, hexadactyly, cleft lip/palate and severe growth and mental retardation. Our patient is interesting for several reasons: lack of any major malformation considered typical for Patau syndrome, the remarkably mild cognitive delay, postnatal low-normal growth and weight, and a pattern of different hemangiomas. To our knowledge only one has been reported with similar skin anomalies without any other organic malformations.

#### 1.P3

### Microdeletion of 15q24 detected by Array-CGH technique, in an Iranian patient with intellectual disability, autism, seizures, and dysmorphism

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15q24 microdeletion syndrome is a rare syndrome with a frequency of about 0.3 % in patients with intellectual disability and about 0.1–0.2 % in patients with autistic symptom disorder. This syndrome was described for the first time in 2007 by Sharp et al. The reported clinical features included growth retardation, intellectual disability, unusual facial characteristics, skeletal and genital anomalies, hypotonia and behavioral problems. Since then 19 individuals have been reported with deletions of different sizes ranging between 1.7 and 6.1 Mbp, detected by array CGH technique. These clinical manifestations are heterogeneous between patients.

We report a 17-year-old boy with multiple clinical features including intellectual disability, autism, unusual craniofacial characteristics and seizures. The high resolution karyotyping was normal 46, XY. The MLPA result for all the subtelomeric regions was also normal. However, array CGH technique using CytoChip international standard cytogenetic array 4× 44 k (v2.0) platform with a resolution of 150–200 kb, revealed a 2.5 Mb deletion of 15q24.1 q25.1.

A genotype phenotype study and comparison with other similar cases will be presented.

#### 1.P4

### Small Deletion of 143 Kb Encompassing Exon 2 of the AUTS2: Rise of a New Microdeletion Syndrome?

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Chromosome microarray analysis is a powerful diagnostic tool and is being used as a first-line approach to detect chromosome imbalances associated with intellectual disability, dysmorphic features and congenital anomalies. This test enables the identification of new

copy number variants (CNVs) and their association with new microdeletion/microduplication syndromes in patients previously without diagnosis.

Here we report the case of a 17 year-old female with severe intellectual disability, absence of speech, microcephaly and congenital abnormalities with a previous normal karyotype performed at a younger age.

Affymetrix CytoScan HD chromosome microarray analysis was performed detecting a 143 Kb deletion at the 7q11.22 breakpoint, encompassing exon 2 of AUTS2 gene: arr[hg19] 7q11.22(69238957-69381975)×1.

The AUTS2 gene has been recently implicated in neurodevelopment and is a candidate gene for numerous neurological disorders. Common clinical features described in patients with deletions in AUTS2 gene include intellectual disability, speech delay and microcephaly, among others. Thus, the CNV identified in our patient explains the phenotype observed.

We compare our patient with other similar reported cases, adding additional value to the phenotype-genotype correlation of deletions in this region. The growing collection of new cases with similar phenotypes, and the observation of this deletion occurring frequently de novo, indicates this CNV as a possible new single gene microdeletion syndrome.

## 1.P5

### Association of structural and numerical anomalies of chromosome 22 in a patient with syndromic intellectual disability.

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Array comparative genomic hybridization CGH is now widely adopted as a first-tier clinical diagnostic test for patients with developmental delay/intellectual disability DD/ID, autism spectrum disorders, and multiple congenital anomalies. Nevertheless, classic karyotyping still has its impact in diagnosing genetic diseases, particularly mosaic cases.

We report on a 30 year old patient with syndromic intellectual disability, a 22q13.2 microdeletion and

mosaic trisomy 22. The patient had the following clinical features: intrauterine growth retardation, mild hypotonia, cryptorchidism, phimosis, facial asymmetry, enophthalmus, mild prognathism, bifid uvula, hypoplastic upper limb phalanges, DD including speech delay, and ID. Whole genome oligonucleotide microarray CGH 105 K (Agilent Technologies) showed a de novo 1 Mb interstitial deletion in 22q13.2, confirmed by fluorescence in situ hybridization in all cells examined. Moreover, 18 % cells had three chromosome 22 signals in addition to the 22q13.2 deletion. G-banded karyotype was done and revealed a trisomy 22 in mosaic.

Almost all 22q13 deletions published so far have been terminal deletions with variable sizes (2.7–6.9 Mb). Very few cases of smaller, interstitial 22q13.2 deletions were reported. In its mosaic form, trisomy 22 is compatible with life, and there are about 20 reports in the literature. It has a variable clinical presentation: growth restriction, dysmorphic features, cardiovascular abnormalities, hemihyperplasia, genitourinary tract anomalies and ID. Neurodevelopmental outcome ranges from normal to severe DD.

Our case points out the role of conventional cytogenetic tools in mosaic cases that could be missed by microarray technology. With the advances of CGH, new microdeletion and microduplication syndromes are being reported. Data is lacking about the interactions of more than one genetic anomaly detected on a same patient, making the genotype/phenotype correlation quite difficult.

## 1.P6

### Mixoploidy combined with aneuploidy in a patient with severe multiple congenital abnormalities and mental retardation

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We report a 10-year-old female patient born to healthy, non-consanguineous parents, presenting with metopic ridge, palatoschisis, dysmorphic features, high-arched eyebrows, multiple congenital contractures, broad